



SLCO1B1 gene

solute carrier organic anion transporter family member 1B1

Normal Function

The *SLCO1B1* gene provides instructions for making a protein called organic anion transporting polypeptide 1B1, or OATP1B1. This protein is found in liver cells; it transports compounds from the blood into the liver so that they can be cleared from the body. For example, the OATP1B1 protein transports bilirubin, which is a yellowish substance that is produced when red blood cells are broken down. In the liver, bilirubin is dissolved in a digestive fluid called bile and then excreted from the body. The OATP1B1 protein also transports certain hormones, toxins, and drugs into the liver for removal from the body. Drugs transported by the OATP1B1 protein include statins, which are used to treat high cholesterol; heart disease medications; certain antibiotics; and some drugs used for the treatment of cancer.

Health Conditions Related to Genetic Changes

Rotor syndrome

Mutations in the *SLCO1B1* gene are involved in Rotor syndrome. This condition is characterized by elevated levels of bilirubin in the blood that can cause yellowing of the skin and whites of the eyes (jaundice). For this condition to occur, individuals must have mutations in the *SLCO1B1* gene and a related gene called *SLCO1B3*. This related gene provides instructions for making a protein called OATP1B3, which has a similar transport function to OATP1B1. In some cases, the condition is caused by a deletion of genetic material that removes parts of both the *SLCO1B1* and *SLCO1B3* genes, so no functional OATP1B1 or OATP1B3 protein is made. Most mutations that cause Rotor syndrome lead to abnormally short, nonfunctioning OATP1B1 and OATP1B3 proteins. Without the function of either transport protein, bilirubin is less efficiently taken up by the liver and cleared from the body. The buildup of this substance leads to jaundice in people with Rotor syndrome.

other disorders

Certain common variations (polymorphisms) in the *SLCO1B1* gene are associated with a reduced ability to process certain drugs, including statins. The most widely studied polymorphism associated with this abnormality, which is found in approximately 15 percent of the population, changes a single protein building block in the OATP1B1 protein: the amino acid valine at position 174 is replaced with the amino acid alanine (written as V174A or *SLCO1B1**5). The protein produced from

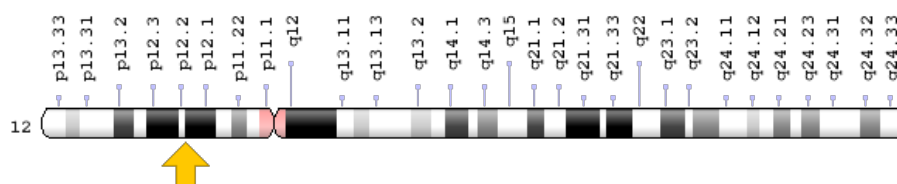
this version of the *SLCO1B1* gene is less able to transport compounds into the liver, which leads to elevated levels of the compounds in the body.

When statins are not efficiently transported into the liver, they build up in the body, and can cause a condition known as statin-induced myopathy. This condition causes fatigue, pain, tenderness, weakness, and cramping in muscles. People with the V174A polymorphism who take statin drugs have an increased risk of developing statin-induced myopathy.

Chromosomal Location

Cytogenetic Location: 12p12.1, which is the short (p) arm of chromosome 12 at position 12.1

Molecular Location: base pairs 21,131,194 to 21,239,796 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HBLRR
- liver-specific organic anion transporter 1
- LST-1
- LST1
- OATP-2
- OATP-C
- OATP1B1
- OATP2
- OATPC
- SLC21A6
- SO1B1_HUMAN
- sodium-independent organic anion-transporting polypeptide 2

- solute carrier family 21 (organic anion transporter), member 6
- solute carrier family 21 member 6
- solute carrier organic anion transporter family, member 1B1

Additional Information & Resources

Educational Resources

- Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals (2009): Pharmacogenomics and Pharmacogenetics
<https://www.ncbi.nlm.nih.gov/books/NBK115549/>

GeneReviews

- Rotor Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK114805>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLCO1B1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER ORGANIC ANION TRANSPORTER FAMILY, MEMBER 1B1
<http://omim.org/entry/604843>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLCO1B1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLCO1B1%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10959
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10959>
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Reviewed: March 2013
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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